TEACHING CASES

Heterochromia

Abstract: A patient was noted to have 2 different eye colours and miosis in her left eye. She ultimately received a diagnosis of congenital Horner syndrome. Determinants of eye colour and possible clinical significance are discussed.

The case: A 35-year-old woman with a hypertensive emergency and confusion presented to the emergency department. Incidentally, we noted that she had 2 different coloured eyes (heterochomia) and miosis of her left eye (Figure 1). The patient reported that her eyes had been different colours since very early in her childhood.

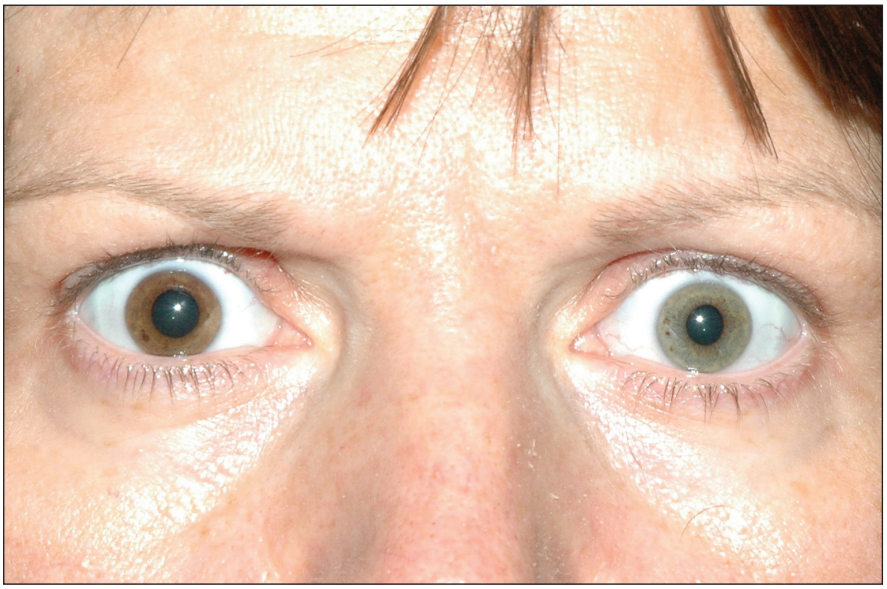

Figure 1: This 35-year-old woman had different coloured eyes since birth. The entire iris of her right eye is brown, and the iris of the left eye is greenish brown. Her left pupil is smaller than the right, which is consistent with the diagnosis of congenital Horner syndrome.

Although some patients have pigment changes involving only 1 segment of the iris (segmental heterochromia or heterochromia iridium),[1] our patient's entire iris was involved (complete heterochromia or heterochromia iridis). Heterochromia iridis is rare, affecting fewer than 200 000 people in the United States.[2] Although uncommon in humans, it is common in some breeds of cats, dogs and horses.

Eye colour is determined by the concentration and distribution of melanin in the iris, with both genetic and physiologic factors affecting determination and maintenance of iris colour. Most human cases of heterochromia are sporadic and benign, and they occur without any detectable underlying abnormality. Congenital heterochromia occurs in a variety of syndromes, including Sturge–Weber syndrome, Waardenburg syndrome and Parry–Romberg syndrome (Table 1). Acquired factors that can lead to heterochromia include ocular trauma, foreign body (ocular siderosis), melanocytic infiltration (diffuse iris nevus or melanoma) and impaired sympathetic tone leading to differential hypo- or hyper-pigmentation of 1 eye. Latanoprost, a glaucoma treatment and a prostaglandin F2α analogue, which was not used by our patient, has also been associated with changing eye colour in up to one-third of people who use the drug for 5 or more years.[3]

Disruption of the sympathetic stimulation of the melanocytes in the superficial stroma of the iris (especially as a child) can lead to heterochromia. Horner syndrome from the unilateral impairment of sympathetic nerves leads to ptosis, miosis, a lag in pupil dilation, enopthalmos (the impression of a sunken eye) and facial anhidrosis (decreased sweating on 1 side of the face). Acquired heterochromia can occur in adults in rare cases as a result of acquired Horner syndrome. In contrast to patients with acquired Horner syndrome, patients with congenital Horner syndrome, such as our patient, often lack several features of the syndrome.

In adults with acquired heterochromia and miosis, Fuchs heterochromic cyclitis and sympathetic heterochromia

Table 1: Differential diagnosis of heterochromia and miosis in an adult

Condition	Clinical features
Waardenburg syndrome	Congenital sensorineural hearing loss, pigmentary abnormalities of iris, hair and skin, craniofacial dysmorphism
Congenital Horner syndrome	Associated with cervical neuroblastoma, carotid dissection, hypoplasia of the internal carotid artery, varicella
Parry–Romberg syndrome	Congenital facial hemiatrophy, severe headache, epilepsy
Fuchs heterochromic iridocyclitis	Acquired visual deterioration, keratic precipitates, cataracts

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must be considered. Unilateral sympathetic nerve lesions such as paravertebral neurilemmoma and neuroblastoma should also be considered. Our patient's clinical presentation was inconsistent with any of these causes. Sympathetic heterochromia was suspected but investigations, including urinary catecholamines and an MIBG (iodine-131-meta-iodobenzylguanidine) scan, did not reveal excess catecholamine secretion or a sympathetic tumour.

The patient's blood pressure was managed with appropriate medication, and she was ultimately discharged from our care with a reversal of her confusion. There was no further follow-up with regard to her eye colour.

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